GENETIC RECESSIVE CONDITIONS IN JAPANESE BLACK & BROWN (WAGYU)

PLEASE NOTE THIS IS A GUIDE ONLY

The scientific terms for the two most common modes of genetic inheritance are: autosomal dominant and autosomal recessive. The term Autosomal means that the condition or trait has a genetic basis (controlled by genes). With cattle breeding a single allele (copy of a gene) comes from each parent for the seven billion genes in the cattle genome.
Autosomal ***dominant*** describes a trait or condition that is expressed when only a single allele (copy of a gene) is present. A practical example of this is the trait of black coat colour. Only one parent is required to input the dominant “black coat colour” allele to have a resultant black coloured calf. Autosomal ***recessive*** describes a trait or condition that is expressed when two alleles are present (one from each parent). Two copies of the recessive allele are required as animals carrying only one copy of the recessive allele do not exhibit the trait (carriers). A practical example of this is the recessive condition CL16. Animals with one copy of the recessive allele (carriers) show no disease symptoms and appear normal. One copy of the recessive allele must be present from both parents to show symptoms of CL16 disease. Two copies of the recessive allele must be present before an animal shows symptoms of the recessive disease (affected animal). ***Carriers*** appear phenotypically (observable characteristics) normal and are indistinguishable from non-carriers unless genetic testing is performed. Normal animals inherit the normal gene from both parents.

PLEASE NOTE that the importance of recessive testing is to identify resultant carriers that may be used as breeding stock. If carrier sires are used as a terminal cross, the impact of harmful recessive conditions is diminished (assuming the breeding herd is a non-carrier or free (F) status) as it would be expected that all progeny is slaughtered.

Five genetic conditions of interest have been identified in the Wagyu breed. The first four listed are generally considered to be lethal disorders and will be **reported** as part of the panel test.
The fifth condition; F11 Deficiency is mild bleeding disorder. Initial research has shown that cattle which are **affected** with the F11 gene show little in the way of easily observable symptoms and the economic impact appears to be minimal. The gene frequency in the Australian Wagyu herd appears to be on par with results for the Japanese Black population. The AWA will continue its research into F11 but, based on the information we have to date F11 testing will **not** be included in the panel test. **The test is however available on request to breeders if they wish to have their animals tested for Factor XI.**

REPORTED CONDITIONS (STANDARD PANEL)

Chediak Higashi Syndrome (**CHS**) status
CHS is a macrophage disorder (a white blood cell that has an important role in the immune response to disease). If cattle have a malfunctioning immune system, this makes them unable to resist bacterial challenge. Blood is slow to coagulate so often the first indicator is unusual umbilical cord haemorrhage at parturition (calving). Cattle with this syndrome often have an unusually pale coat colour.
PHOTOGRAPH – Affected newborn calf with a pale coat

Spherocystosis (**B3**) status
This is a disorder of the surface membrane of the erythrocyte (red blood cells). The protein from the B3 gene makes up the basic structure of the erythrocyte. Cattle that are homozygous (have two copies of the recessive allele) have pernicious anaemia (bleeding caused by the abnormal red blood cells). Death normally occurs within the first 7 days after birth. Some cases live to adulthood but there is a severe retardation in growth.
PHOTOGRAPHS - Icterus in the eyes &, anaemic calf @ 5m (90 kg lwt)

Claudin 16 deficiency (**CL16**) status (Type 1)
Claudin 16 or RTD(Renal tubular dysplasia) is a gene disorder on chromosome 1 and causes kidney failure (chronic interstitial nephritis (CIN), often with zonal fibrosis or excess of fibrous connective tissue). There are 2 types of the deletion in this gene that can cause CIN. This disorder results in terminal kidney failure and the onset can occur any time from late adolescence. Cattle are unlikely to live more than 6 years.
PHOTOGRAPH - Elongated Hooves on an affected CL16

Factor 13 deficiency (**F13**) status
This gene contributes to fibrin stability which is an integral part of the blood coagulation pathway (blood clotting ability). Disorders in this gene cause severe haemorrhage (bleeding). In calves haemorrhage is particularly likely to occur in the hindquarters causing blood to pool and stagnate under the hide. In adult cattle any minor trauma (such as hitting the animal) can cause major haemorrhage at the trauma site.
PHOTOGRAPH - Navel of an affected F13 newborn

NB Photo source – Color Atlas of Bovine Congenital Defects (Ed. K.Hamana: 2006)

ADDITIONAL TEST (IF REQUIRED)

Factor XI deficiency (**F11**) status (usually non-lethal)
Factor XI (F11) is a plasma protein that participates in the formation of blood clots. Factor XI deficiency is an autosomal disorder that is associated with mild bleeding in Wagyu. Affected animals show prolonged bleeding time and abnormal plasma coagulation after trauma or surgical procedures such as castration or dehorning.
PHOTOGRAPH – Affected F11 Female exhibiting no harmful symptoms

Recessive gene status reporting

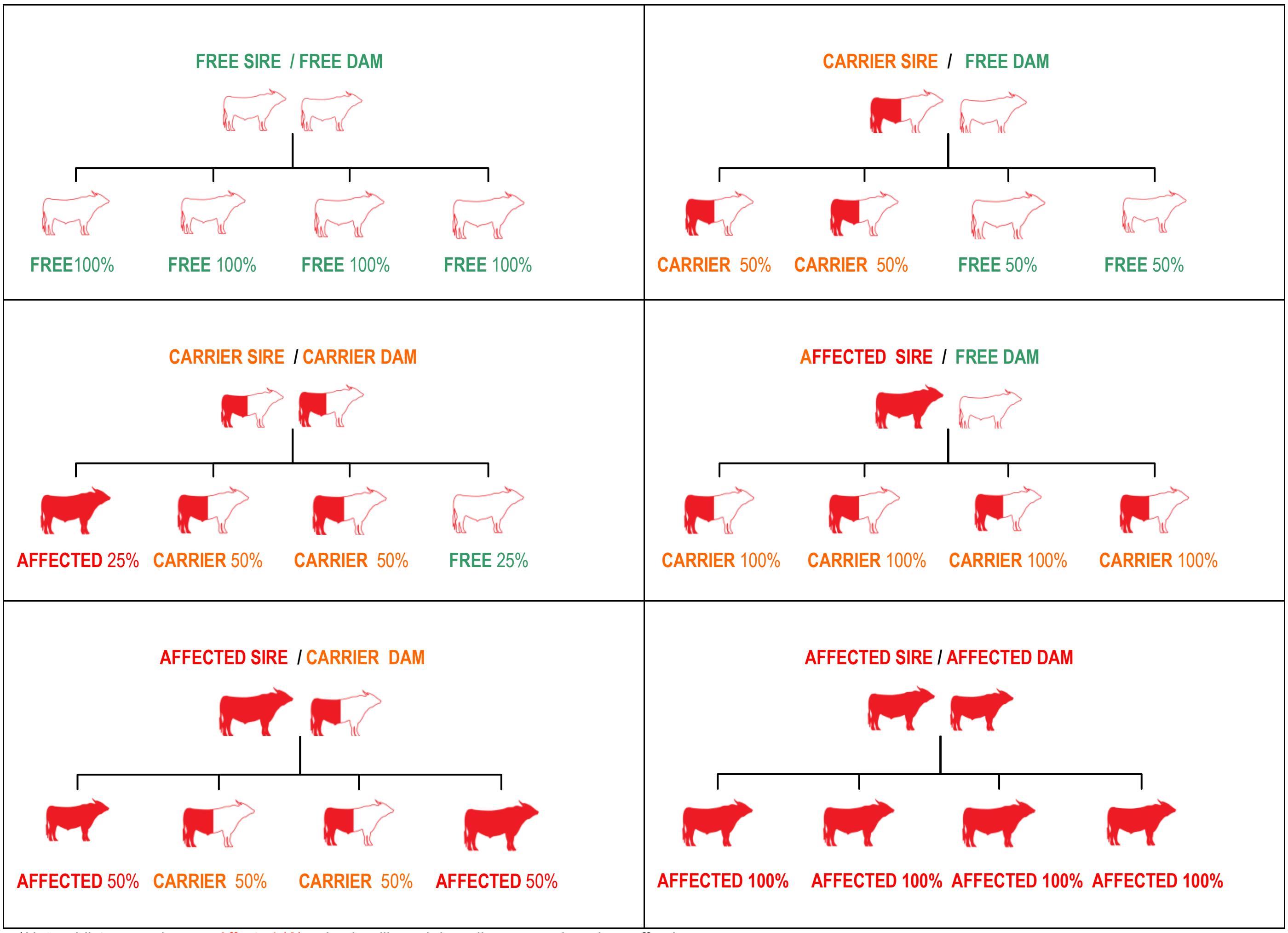
DNA tests will report the status of animal using three (3) categories:

F	**Free** indicates that the sample submitted for this animal **has been tested** and found to be **clear** of the causative mutation (abnormal gene) responsible for an identified autosomal condition. This animal is homozygous free, meaning that it has two copies of the normal allele.
C	**Carrier** indicates that the sample submitted for this animal **has been tested** and found to have one copy of the causative mutation responsible for an identified autosomal condition. This animal is heterozygous for the mutation, meaning that it has one abnormal allele and one normal allele. This animal could pass the mutation (abnormal gene) to approximately half of its progeny.
A	**Affected** calves are rarely tested. They are homozygous for the mutation responsible for the autosomal condition and have two copies of the abnormal variant of the gene.

What does this mean for management of the genetic conditions?

As one gene comes from each parent; expected progeny results of breeding different genetic condition status sires & dams are listed in table format below *Figure 1* (**Free (F)**, **Carrier (C)** & **Affected (A)** status);

Figure 1 - example gene combinations in autosomal recessive parent/s and the resultant offspring.



**Note whilst unusual, some Affected (A) animals will reach breeding age and produce offspring.*

Dealing with autosomal recessive conditions

Unlike diseases that are the result of pathogenic agents such as bacteria or viruses that may result in heavy losses of animals and major de-contamination procedures, genetic conditions are not contagious and CANNOT be transmitted merely by animal-to-animal contact. Control of genetic conditions requires good management practices. These are developed based on the aims and objectives of each enterprise.

Figure 2 – Likely scenario of gene combinations using a carrier status sire & free status dam herd for the first generation and the resultant female offspring being bred for the next three (3) generations to a free status sire and on the fifth(5) generation a carrier status sire is re-introduced into the breeding program. Although no clinical signs have been seen for four generations, reintroduction of carrier status sire can have an economic impact with affected status animals appearing in resultant progeny in the fifth generation.

Generation 1(G1) *Introduce a carrier sire to free dam*

Generation 2(G2) *by joining a free sire to the retained heifers from generation1 knowing that 50% are carrier status*

Generation 3(G3) *by joining a free sire to the retained heifers from generation 2 knowing that 25% are carrier status*

Generation 4(G4) *by joining a free sire to the retained heifers from generation 3 knowing that 12.5% of are carrier status*

***Generation 5(G5)** *Re-introduce a carrier sire to the retained heifers from generation 4 knowing that 6.25% of are carrier status*

Management Options for autosomal recessive conditions

Many management options exist for the control of genetic conditions and these are formulated based on the individual needs and requirements of each enterprise. Some of these include:

- Test all animals and remove carriers.
- Test all animals and use carriers ONLY in terminal breeding programmes.
- Test sires and only use free bulls for breeding. This will eliminate affected progeny and decrease the number of carriers over time.
- These are just three examples of management practices that can be used exclusively or in combination. Please note this is NOT an exhaustive list of the options available.

What about cross-breeds?

Autosomal recessive conditions can be perpetuated in cross-breeds. First crosses are unlikely to produce any affected progeny (unless both breeds have the disease), but there is a risk of increasing the prevalence of the defective gene in the carrier state. Subsequent inter-breeding of cross-bred animals is likely to produce additional carriers and affected cases.

Can you retain superior production genetics from carrier animals?

YES! Carrier sires and dams can be used for breeding. However, follow-up testing of all progeny would be essential to develop future breeding strategies with that progeny. Expect a minimum of 50% of their progeny to be carriers. These carriers can be identified by DNA testing. They may be culled, or used in a separate breeding programme with animals of identified negative or free status. The Negative status progeny can be used to perpetuate phenotypically and/or quantitatively selected superior production traits. Continued breeding by joining TWO carrier animals is not recommended due to the risk of producing approximately 25% affected animals.

Q & A

Q: What is a Free (F)?
A: Zero copies of the mutant recessive allele – The animal is completely **free** of the genes that cause the particular disease.

Q: What is the effect on use of an F status animal in a breeding program?
A: No spread of recessive inherited disease from F animal

Q: What are the clinical signs of an F animal?
*A: **CLINICALLY NORMAL** – NO DISEASE SYMPTOMS*

Q: What is a Carrier (C)?
A: Carriers have one copy of the mutant recessive allele

Q: What is the effect on use of a C status animal in a breeding program?
A: 50% of the progeny will receive one copy of the mutant gene. If the other parent is either a carrier or affected the resultant progeny can also be affected.

Q: What are the clinical signs of a C animal?
*A: **CLINICALLY NORMAL** – NO DISEASE SYMPTOMS*

Q: What is an Affected (A)?
A: Affected animals have two copies of the mutant recessive allele

Q: What is the effect on use of an A status animal in a breeding program?
A: *100% progeny will receive 1 mutant gene from the affected parent. If the other parent is either a carrier or affected the resultant progeny can also be affected.*

Q: What are the clinical signs of an A animal
*A: **CLINICALLY AFFECTED** – **DISEASE SYMPTOMS DISPLAYED***

Costs

AWA has negotiated a fee structure with the Elizabeth Macarthur Agricultural Institute (EMAI) to perform the test panel. The 2010 pricing is $140 ex. GST for a four(4) panel test (if DNA is held at UQ AGL.) If profile is required and DNA type is needed from hair, semen or blood POA

*Breeder request to test for **Factor XI (F11)**, then an additional fee of $35 fee is charged and a five(5) panel test will be conducted for $175 ex GST.*

All animals must be registered with the AWA and have PV on record. All initial inquires / testing need to be directed to Michael Beattie on 02 6773 3355 or office@wagyu.org.au

The Australian Wagyu Association (AWA) web database will display the recessive condition results from tested animals in mid April, 2010. This information can be accessed by conducting a "Database Search" from the AWA website and viewing reported results on the animal details page under "Gene Tests". The status of *untested* animals is subject to change. Links to the AWA recessive testing application form along with supporting scientific publications on recessive conditions in Wagyu will be available via AWA website.

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